



## ATXN1 gene

ataxin 1

### Normal Function

The *ATXN1* gene provides instructions for making a protein called ataxin-1. This protein is found throughout the body, but its function is unknown. Within cells, ataxin-1 is located in the nucleus. Researchers believe that ataxin-1 may be involved in regulating various aspects of producing proteins, including the first stage of protein production (transcription) and processing RNA, a chemical cousin of DNA.

One region of the *ATXN1* gene contains a DNA segment known as a CAG trinucleotide repeat. This segment is made up of a series of three DNA building blocks (cytosine, adenine, and guanine) that appear multiple times in a row. Normally, the CAG segment is repeated 4 to 39 times within the gene.

### Health Conditions Related to Genetic Changes

#### spinocerebellar ataxia type 1

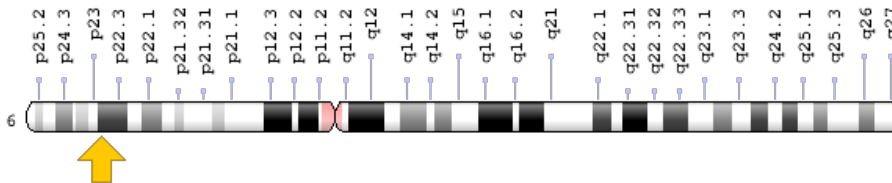
Spinocerebellar ataxia type 1 (SCA1) is a condition characterized by progressive problems with movement. SCA1 results from a mutation in the *ATXN1* gene known as a trinucleotide repeat expansion. This mutation increases the length of the repeated CAG segment in the *ATXN1* gene. People with SCA1 have from 40 to more than 80 CAG repeats in most cells.

The expanded CAG segment leads to the production of an abnormally long version of the ataxin-1 protein that folds into the wrong 3-dimensional shape. This abnormal protein clusters with other proteins to form clumps (aggregates) within the nucleus of cells. These aggregates prevent the ataxin-1 protein from functioning normally, which damages cells. For reasons that are unclear, aggregates of ataxin-1 are found only in the brain and spinal cord (central nervous system). Certain brain cells called Purkinje cells seem to be particularly sensitive to the accumulation of these aggregates. Purkinje cells are located in the part of the brain that coordinates movement (cerebellum) and are involved in chemical signaling between nerve cells (neurons). The accumulation of protein aggregates in the cells of the central nervous system, particularly in Purkinje cells, leads to cell death. Over time, the loss of these cells causes the movement problems characteristic of SCA1.

## Chromosomal Location

Cytogenetic Location: 6p22.3, which is the short (p) arm of chromosome 6 at position 22.3

Molecular Location: base pairs 16,299,112 to 16,761,490 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ataxin-1
- ATX1
- ATX1\_HUMAN
- SCA1

## Additional Information & Resources

### Educational Resources

- Genes and Disease: Spinocerebellar Ataxia  
<https://www.ncbi.nlm.nih.gov/books/NBK22234/>
- Washington University, St. Louis: Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/ataxia/domatax.html#sca1>

### GeneReviews

- Spinocerebellar Ataxia Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1184>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ATXN1%5BTIAB%5D%29+OR+%28ataxin+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- ATAXIN 1  
<http://omim.org/entry/601556>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ATXN1.html](http://atlasgeneticsoncology.org/Genes/GC_ATXN1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ATXN1%5Bgene%5D>
- HGNC Gene Family: Ataxins  
<http://www.genenames.org/cgi-bin/genefamilies/set/411>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=10548](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10548)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6310>
- UniProt  
<http://www.uniprot.org/uniprot/P54253>

## **Sources for This Summary**

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